



TAT gene

tyrosine aminotransferase

Normal Function

The *TAT* gene provides instructions for making a liver enzyme called tyrosine aminotransferase. This enzyme is the first in a series of five enzymes that work to break down the amino acid tyrosine, a protein building block found in many foods. Specifically, tyrosine aminotransferase converts tyrosine into a byproduct called 4-hydroxyphenylpyruvate. Continuing the process, 4-hydroxyphenylpyruvate is further broken down and ultimately smaller molecules are produced that are either excreted by the kidneys or used to produce energy or make other substances in the body.

Health Conditions Related to Genetic Changes

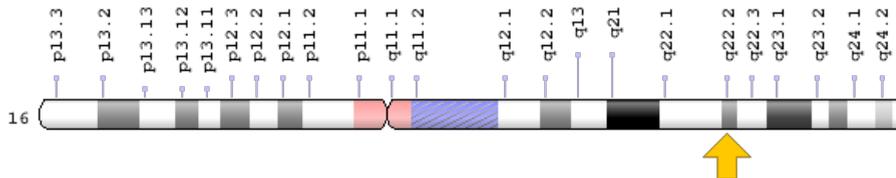
tyrosinemia

At least 22 *TAT* gene mutations have been found to cause tyrosinemia type II. This condition often affects the eyes, skin, and mental development. Most of these mutations change single DNA building blocks (base pairs) within the *TAT* gene. Research suggests that the altered *TAT* gene produces a tyrosine aminotransferase enzyme with reduced activity. Other mutations delete all or part of the *TAT* gene, eliminating enzyme activity. As a result of these mutations, tyrosine is not properly broken down. Tyrosine levels are elevated and some tyrosine is converted into other molecules that may be toxic to cells. It is unclear how impaired break down of tyrosine leads to the skin, eye, and intellectual problems that characterize tyrosinemia type II.

Chromosomal Location

Cytogenetic Location: 16q22.2, which is the long (q) arm of chromosome 16 at position 22.2

Molecular Location: base pairs 71,566,851 to 71,577,095 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ATTY_HUMAN
- L-tyrosine:2-oxoglutarate aminotransferase
- tyrosine transaminase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Phenylalanine and Tyrosine Degradation (figure)
<https://www.ncbi.nlm.nih.gov/books/NBK22453/figure/A3256/?report=objectonly>

GeneReviews

- Tyrosinemia Type I
<https://www.ncbi.nlm.nih.gov/books/NBK1515>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28tyrosine+aminotransferase%5BTIAB%5D%29+NOT+%28glucocorticoid%5BTIAB%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- TYROSINE AMINOTRANSFERASE
<http://omim.org/entry/613018>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TAT.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TAT%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11573
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6898>
- UniProt
<http://www.uniprot.org/uniprot/P17735>

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